



FAS gene

Fas cell surface death receptor

Normal Function

The *FAS* gene provides instructions for making a protein that is involved in signaling. Three FAS proteins group together to form a structure called a trimer, which then interacts with other molecules to perform its signaling function. This signaling initiates a process called a caspase cascade. The caspase cascade is a series of steps that results in the self-destruction of cells (apoptosis) when they are not needed.

Health Conditions Related to Genetic Changes

autoimmune lymphoproliferative syndrome

More than 100 mutations in the *FAS* gene have been identified in people with a disorder of the immune system called autoimmune lymphoproliferative syndrome (ALPS). ALPS is characterized by the production of an abnormally large number of immune system cells (lymphocytes), resulting in enlargement of the lymph nodes (lymphadenopathy), the liver (hepatomegaly), and the spleen (splenomegaly). Autoimmune disorders, in which the immune system malfunctions and attacks the body's own tissues and organs, are also common in ALPS. People with ALPS have an increased risk of developing cancer of the immune system cells (lymphoma) and may also be at increased risk of developing other cancers.

When the immune system is activated to fight an infection, large numbers of lymphocytes are produced. Normally, these lymphocytes undergo apoptosis when they are no longer required. *FAS* gene mutations result in an abnormal trimer that interferes with the initiation of apoptosis. Excess lymphocytes accumulate in the body's tissues and organs and often begin attacking them, leading to autoimmune disorders. Interference with apoptosis allows cells to multiply without control, leading to the lymphomas and other cancers that occur in people with this disorder.

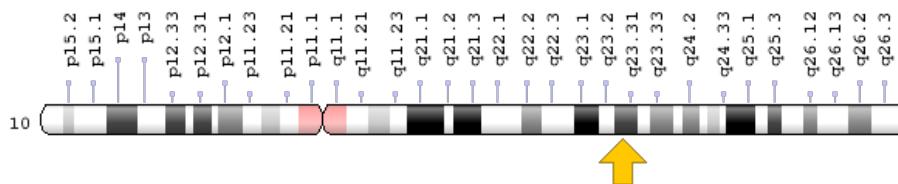
cancers

Studies have associated certain *FAS* gene variations with increased risk of developing cancer, including cancers of the lung, breast, and esophagus. Researchers believe that these variations may affect the signaling that initiates apoptosis, increasing the risk that cells will multiply out of control and result in cancer.

Chromosomal Location

Cytogenetic Location: 10q23.31, which is the long (q) arm of chromosome 10 at position 23.31

Molecular Location: base pairs 88,968,429 to 89,017,061 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- APO-1
- apo-1 antigen
- APO-1 cell surface antigen
- apoptosis antigen 1
- apoptosis-mediating surface antigen FAS
- APT1
- CD95
- CD95 antigen
- Fas (TNF receptor superfamily, member 6)
- Fas AMA
- Fas antigen
- FAS1
- FASLG receptor
- TNFRSF6
- TNR6_HUMAN
- tumor necrosis factor receptor superfamily member 6

Additional Information & Resources

Educational Resources

- Immunology (fifth edition, 2001): Programmed Cell Death of Activated Lymphocytes is Triggered Mainly Through the Receptor Fas
<https://www.ncbi.nlm.nih.gov/books/NBK27094/#A718>
- National Institute of Allergy and Infectious Diseases (NIAID): ALPS Cause
<https://www.niaid.nih.gov/diseases-conditions/autoimmune-lymphoproliferative-syndrome-causes>

GeneReviews

- Autoimmune Lymphoproliferative Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1108>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FAS%5BTI%5D%29+OR+%28Fas%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

OMIM

- FAS CELL SURFACE DEATH RECEPTOR
<http://omim.org/entry/134637>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/FASID207ch10q23.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FAS%5Bgene%5D>
- HGNC Gene Family: CD molecules
<http://www.genenames.org/cgi-bin/genefamilies/set/471>
- HGNC Gene Family: Death inducing signaling complex
<http://www.genenames.org/cgi-bin/genefamilies/set/1342>
- HGNC Gene Family: Tumor necrosis factor receptor superfamily
<http://www.genenames.org/cgi-bin/genefamilies/set/782>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11920

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/355>
- UniProt
<http://www.uniprot.org/uniprot/P25445>

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Reviewed: July 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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